

(12) INTERNATIONAL APPLICATION PUBLISHED UNDER THE PATENT COOPERATION TREATY (PCT)

(19) World Intellectual Property Organization International Bureau



(43) International Publication Date  
6 January 2005 (06.01.2005)

PCT

(10) International Publication Number  
**WO 2005/001113 A3**

(51) International Patent Classification<sup>7</sup>: C12Q 1/68, C12P 19/34, C07H 21/04

(21) International Application Number:

PCT/US2004/020464

(22) International Filing Date: 25 June 2004 (25.06.2004)

(25) Filing Language:

English

(26) Publication Language:

English

(30) Priority Data:

60/483,352 27 June 2003 (27.06.2003) US

(71) Applicants (for all designated States except US): THOMAS JEFFERSON UNIVERSITY [US/US]; 1020 Walnut Street, Philadelphia, PA 19107 (US). THE TRUSTEES OF THE UNIVERSITY OF PENNSYLVANIA [US/US]; 3160 Chestnut Street, Suite 200, Philadelphia, PA 19104 (US).

(72) Inventors; and

(75) Inventors/Applicants (for US only): DELGROSSO, Kathleen [US/US]; 205 Pennsylvania Avenue, Aldan, PA 19018 (US). FORTINA, Paolo [US/US]; 2020 Walnut Street, Apt. 10-E, Philadelphia, PA 19130 (US). GRAVES, David [US/US]; 221 Highland Avenue, Devon, PA 19333 (US). SURREY, Saul [US/US]; 5 Grouse Trail, Chadds Ford, PA 19317 (US).

(74) Agent: MONACO, Daniel, A.; Drinker Biddle & Reath, LLP, One Logan Square, 18th and Cherry Streets, Philadelphia, PA 19103-6996 (US).

(81) Designated States (unless otherwise indicated, for every kind of national protection available): AE, AG, AL, AM, AT, AU, AZ, BA, BB, BG, BR, BW, BY, BZ, CA, CH, CN, CO, CR, CU, CZ, DE, DK, DM, DZ, EC, EE, EG, ES, FI, GB, GD, GE, GH, GM, HR, HU, ID, IL, IN, IS, JP, KE, KG, KP, KR, KZ, LC, LK, LR, LS, LT, LU, LV, MA, MD, MG, MK, MN, MW, MX, MZ, NA, NI, NO, NZ, OM, PG, PH, PL, PT, RO, RU, SC, SD, SE, SG, SK, SL, SY, TJ, TM, TN, TR, TT, TZ, UA, UG, US, UZ, VC, VN, YU, ZA, ZM, ZW.

(84) Designated States (unless otherwise indicated, for every kind of regional protection available): ARIPO (BW, GH, GM, KE, LS, MW, MZ, NA, SD, SL, SZ, TZ, UG, ZM, ZW), Eurasian (AM, AZ, BY, KG, KZ, MD, RU, TJ, TM), European (AT, BE, BG, CH, CY, CZ, DE, DK, EE, ES, FI, FR, GB, GR, HU, IE, IT, LU, MC, NL, PL, PT, RO, SE, SI, SK, TR), OAPI (BF, BJ, CF, CG, CI, CM, GA, GN, GQ, GW, ML, MR, NE, SN, TD, TG).

Published:

— with international search report

(88) Date of publication of the international search report:  
28 April 2005

For two-letter codes and other abbreviations, refer to the "Guidance Notes on Codes and Abbreviations" appearing at the beginning of each regular issue of the PCT Gazette.

WO 2005/001113 A3

(54) Title: METHODS FOR DETECTING NUCLEIC ACID VARIATIONS

(57) Abstract: The invention provides methods and diagnostic test kits for detecting target nucleic acid sequence variations in a sample. In particular, a plurality of oligonucleotide probe sets is provided. Each set has a target specific portion and a barcode. The target specific portions of the probes are suitable for ligation together when hybridized adjacent to one another on a corresponding target polynucleotide. The ligated product contains a barcode that binds to a probe attached to a substrate and hence facilitate the capture of the ligated product on the solid support. Detection is carried out with a gold nanoparticle-attached barcode that hybridizes with the barcode on the ligated product.